



SLC16A2 gene

solute carrier family 16 member 2

Normal Function

The *SLC16A2* gene (also known as *MCT8*) provides instructions for making a protein that plays a critical role in nervous system development. This protein transports a particular hormone into nerve cells in the developing brain. This hormone, called triiodothyronine or T3, is produced by the thyroid (a butterfly-shaped gland in the lower neck). Once inside a nerve cell, T3 interacts with receptors in the nucleus that turn specific genes on or off. The activity of this hormone appears to be critical for the maturation of nerve cells, the movement of these cells to their proper locations (cell migration), and the formation of specialized cell outgrowths called dendrites. T3 may also play a role in the development of synapses, which are junctions between nerve cells where cell-to-cell communication occurs.

In addition to the nervous system, T3 is produced in the liver, kidney, heart, and several other tissues. T3 and other forms of thyroid hormone help regulate the development of many organs and control the rate of chemical reactions in the body (metabolism).

Health Conditions Related to Genetic Changes

Allan-Herndon-Dudley syndrome

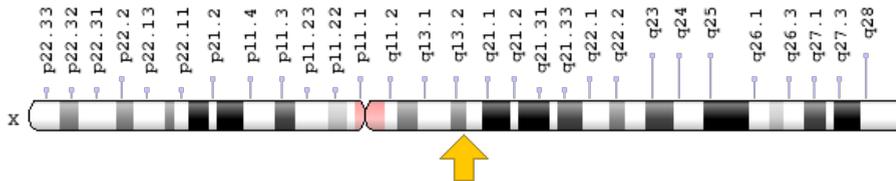
At least a dozen mutations in the *SLC16A2* gene have been identified in people with the characteristic features of Allan-Herndon-Dudley syndrome. Some of these mutations insert or delete genetic material in the gene. Other mutations change single protein building blocks (amino acids) used to make the *SLC16A2* protein. All of these genetic changes alter the structure and function of this protein, preventing it from transporting T3 into nerve cells effectively. A lack of this critical hormone in certain parts of the brain disrupts normal brain development, resulting in intellectual disability and problems with movement.

If T3 is not taken up by nerve cells, excess amounts of this hormone continue to circulate in the bloodstream. An increase in circulating T3 may be toxic to some organs, such as the liver. Researchers are working to determine whether increased T3 levels in the body contribute to the signs and symptoms of Allan-Herndon-Dudley syndrome.

Chromosomal Location

Cytogenetic Location: Xq13.2, which is the long (q) arm of the X chromosome at position 13.2

Molecular Location: base pairs 74,421,493 to 74,533,929 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DXS128E
- MCT8
- monocarboxylate transporter 8
- MOT8_HUMAN
- solute carrier family 16 (monocarboxylic acid transporters), member 2
- solute carrier family 16, member 2 (monocarboxylic acid transporter 8)
- solute carrier family 16, member 2 (thyroid hormone transporter)
- X-linked PEST-containing transporter
- XPCT

Additional Information & Resources

Educational Resources

- Endocrinology: An Integrated Approach (first edition, 2001): Actions of Thyroid Hormones
<https://www.ncbi.nlm.nih.gov/books/NBK28/#A280>

GeneReviews

- MCT8-Specific Thyroid Hormone Cell-Membrane Transporter Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK26373>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC16A2%5BTIAB%5D%29+OR+%28%28AHDS%5BTIAB%5D%29+OR+%28MCT8%5BTIAB%5D%29+OR+%28monocarboxylate+transporter+8%5BTIAB%5D%29+OR+%28X-linked+PEST-containing+transporter%5BTIAB%5D%29+OR+%28XPCT%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 16 (MONOCARBOXYLIC ACID TRANSPORTER), MEMBER 2
<http://omim.org/entry/300095>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC16A2%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Family: X-linked mental retardation
<http://www.genenames.org/cgi-bin/genefamilies/set/103>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10923
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6567>
- UniProt
<http://www.uniprot.org/uniprot/P36021>

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